PO-0091 5 YEAR OLD GIRL WITH DISPROPORTIONATE SHORT STATURE WITH FAILURE TO THRIVE AND DYSMORPHISM: RARE CASE OF SCHEIE SYNDROME

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Background Hurler-Scheie Syndrome is an autosomal recessive mucopolysaccharidois resulting in reduced activity of α-L-iduronidase with accumulation of heparin and dermatan sulphate. Scheie syndrome represents less severely affected form with varied clinical features.

Case Report 5 year female, first born child of 2nd degree consanguineous Egyptian parents, referred for short stature presented with following features.

On examination:

Length: 88 cm; Upper: Lower segment=1:1.3; weight: 12.3 kg; Head circumference: 51 cm

Protuberant abdomen, mild proptosis, enlarged skull, prominent forehead, low set ears, simian crease, clawed hand, mild kyphosis, strabismus, normal IQ, hepatomegaly.

Investigation: USG: hepatomegaly, Karyotyping normal Mild delay bone age; Elevated TSH, low T4; tissue transglutaminase normal; sweat chloride test normal; ammonia, lactate normal; growth hormone assay normal; Deficient enzyme in fibroblast confirmed diagnosis of Scheie syndrome

Discussion Scheie syndrome is an autosomal recessive, rare lysosomal storage disease, with skeletal deformities and motor delay; described first in 1972; caused by mutations in *IDUA* gene (4p16.3) leading to partial deficiency in alpha-L-iduronidase enzyme and lysosomal accumulation of dermatan and heparan sulfate. Genetic testing is available. Antenatal diagnosis done by measurement of enzymatic activity in chorionic villus/ amniocytes and by genetic testing (if disease-causing mutation is known). Genetic counselling is recommended. Management is multidisciplinary including physiotherapy (to maintain range of movement); bone marrow or umbilical cord blood transplant (to preserve neurocognition, improve somatic disease and increase survival). Enzyme replacement therapy slows disease progression.

Abstract PO-0091 Figure 1

Conclusion Scheie syndrome should be considered in differential diagnosis of disproportionate short stature with dysmorphism, failure to thrive with normal IQ.

PO-0092 GC-MS IN DIAGNOSIS OF BEHAVIOURAL ABNORMALITIES AND DYSMORPHISM : A CASE SERIES

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Background Several metabolic disorders can present with behavioural abnormalities or autism. There may or maynot be associated seizures or acute metabolic decompensation. Gas chromatography and mass spectrometry (GC-MS) is a modality for non-invasive testing for some of these disorders, especially organic acidurias. Children with dysmorphism, malformations and metabolic disorders are followed up in Genetic Clinic.

Aims We present here 7 cases of metabolic disorders diagnosed on GC-MS testing on urine and on follow up in the Genetic Clinic.

Methods The cases of suspected metabolic disorders were worked up through the outpatient clinic or during admission in Genetic ward. Patients seen in l1 year (Jan. to Dec. 2014) were included in the study.

Results There were 4 cases of methylmalonic acidemia (MMA), one case each of 3-methylglutaconic aciduria, didhydrolipoyldehydrogenase (DLD) deficiency and MMA with homocystinuria. The cases with MMA presented with developmental delay, hyperactivity and behavioural abnormalies. There was subtle dysmorphism on clinical evaluation. The child with MMA with homocystinuria presented with mental retardation and prominent forehead. He was admitted earlier with an episode of DVT and detailed evaluation led to the diagnosis. The child with 3-methyl-glutaconic aciduria presented with behavioural abnormalities.

Conclusions MMA was found to be a commoner metabolic disorder and treatable. A high index of suspicion is needed in diagnosis and an early diagnosis can improve the outcomes in patients with metabolic disorder.

PO-0093 FAMILIAL PURE GONADAL DYSGENESIS

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Introduction Gonadal dysgenesis in females which would be presented with 46 XX is characterised by the presence of primary amenorrhea with or without normal secondary sexual characteristics. It is commonly described without normal secondary sexual characteristics such as breast development. In this study, we aimed to report a familial pure gonadal dysgenesis with and without normal secondary sexual characteristics.

Case presentations In a family consisting 9 siblings of non-consanguineous parents, three sisters (age 22, 17 and 13 years) presented with complaints of amenorrhoea. The 13 years old girl mentioned no secondary characteristics of puberty. The 17 years old girl showed secondary characteristics of puberty without previous history of hormone therapy. Also, the 22 years old girl showed secondary characteristics and clinicians recommended hormone therapy after marriage for her infertility and even delivered a baby. The elder two sisters had a normal female

phenotype and the youngest had amenorrhea with no breast development (B1) and pubic hair. Furthermore, according to physical examination, deafness was not mentioned.

Conclusions According to results, it seems that clinicians should consider different presentation for pure gonadal dysgenesis with familial pattern and further evaluation is needed in malignant degeneration of the gonadal dysgenesis in the patients with 46, XX PGD.

PO-0094 ANTHROPOMETRIC CHARACTERISTICS AND BONE MINERAL DENSITY IN PATIENTS WITH PHENYLKETONURIA

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Background Phenylketonuria (PKU) treatment requires a diet restricted from natural proteins and supplemented with phenylalanine (Phe)-free L-amino acid mixtures. Growth impairment and compromised bone mass have been described.

This study aims to evaluate anthropometric characteristics and bone mineral density (BMD) in a cohort of PKU patients.

Methods We conducted a retrospective longitudinal study collecting anthropometric characteristics (weight, height, body mass index (BMI) and BMD every 6 months from birth to 12 years of age in 34patients with diet restrictions.

We compared the data results, expressed as z-scores, with the general population, as well as between patients with Phe <360 mmol/ml (optimal) and patients with >360 mmol/ml.

Results Our PKU patients are shorter than the reference population; the sample mean was below z–score=0. Weight was comparable to that of the reference population and BMI had a tendency to be over the population mean.

Growth impairment in PHA-deficiency is not related to plasma Phe concentration at birth but might be related to its levels throughout the follow up; patients with <360 mmol/ml were shorter.

BMD was below the population mean in all cases (52% osteopenia).

Conclusions PKU children in our study have a below than average height. Weight is consistent with the population average and BMI tends to be above it.

Height seems more affected among those patients with better metabolic control.

BMD is below the population mean. These data do not vary depending on the levels of Phe at diagnosis, but by the phenylalaninemia during growth.

Further studies are needed to investigate the effect of diet restriction in PKU.

PO-0095 EARLY PROGRAMMING OF AEROBIC AND NEUROMUSCULAIR FITNESS AT PRIMARY SCHOOL AGE. THE ABCD-STUDY

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Background and aims Low birth weight and accelerated postnatal growth are associated with adult cardiovascular disease. In this perspective body composition and obesity can result from a disturbed energy balance due to early reprogramming of energy intake and expenditure. We hypothesise that low birth weight and accelerated growth may predispose ("program") reduced physical fitness at 8–9 years of age.

Methods Aerobic fitness was measured using a 20 metre multistage shuttle run test (20m-MSRT) and neuromuscular fitness using the standing broad jump (SBJ) test and handgrip strength test was measured in 194 children (104 boys) of Dutch ethnicity at mean age 8.6 years in a prospective birth cohort.

Results Subjects with low birth weight and accelerated infant growth reached mean (±SD) 20m-MSRT levels of 3,9 which was significantly lower than (1) normal birth weight and normal infant growth (2) low birth weight and normal infant growth and (3) normal birth weight and accelerated infant growth groups (all $p < 0.01$). Low birth weight subjects had mean grip strength of 12.3 kg (±3.0), which was significantly lower than normal weight subjects with no effect of infant growth on this relationship. There was no association of birth weight or infant growth with grip strength or SBJ.

Conclusions Low birth weight with accelerated infant weight gain was associated with diminished aerobic fitness. Higher birth weight was associated with increased neuromuscular fitness. These early changes may explain increased susceptibility to obesity and related risk factors in low birth weight and early growth accelerated children.

PO-0096 CLINICAL EFFECTIVENESS OF IDURSULFASE IN BOYS AGED 0–5 YEARS WITH HUNTER SYNDROME: 3-YEAR DATA FROM THE HUNTER OUTCOME SURVEY

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Background and aims Symptoms of Hunter syndrome typically become apparent at 2–4 years of age. Previous analyses have demonstrated improvements in certain clinical measures in young patients receiving idursulfase (Shire); however, data on long-term idursulfase use in these patients remain limited. This analysis used data available in the Hunter Outcome Survey (HOS), a global, observational registry sponsored by Shire, to investigate long-term effectiveness of idursulfase in boys with Hunter syndrome aged 0–5 years.

Methods As of January 2014, 260/564 males followed prospectively in HOS had received ≥1 idursulfase infusion (excluding those who had received a bone marrow transplant or were enrolled in the TKT018/TKT024 clinical trials), were aged 0–5 years at treatment initiation and were included in the analysis. Median age at first treatment was 3.5 years; median treatment duration was 41.6 months. Clinical measures recorded in HOS at annual timepoints over 3 years were compared with baseline values.

Results Median urinary glycosaminoglycan (uGAG) levels, liver size and left ventricular mass index had improved at all yearly